TruGenome Undiagnosed Disease Test

Helping you and your patients find answers when faced with the challenge of an undiagnosed disease.

By their very nature, undiagnosed diseases are observed in only a small percentage of the population. This rarity makes it difficult for current molecular tests to find the root genetic cause, as they often target the more commonly found genetic variants. For rare diseases, it may be necessary to look at the whole genome to find the causative variant. Today, more than ever before, new discoveries and technologies are increasing access to our DNA, helping us make sense of the information it contains.

Next-generation sequencing (NGS) technology is leading the genetic information revolution. In fact, NGS technologies are proving to be successful in finding the causes of rare genetic diseases[1,2]. Taking advantage of these developments, the TruGenome Undiagnosed Disease Test sequences the whole genome, enabling a single test to provide you with a deeper look into your patient's genome. This gives you a new opportunity to potentially find answers more efficiently and cost-effectively. By sequencing the whole genome, you can review your patient's DNA in light of current information and reanalyze as new discoveries are made. Working with a patient presenting with an undiagnosed disease can be a challenging experience for all involved. The TruGenome Undiagnosed Disease Test can offer you and your patient hope in these uncertain circumstances.

What is the TruGenome Undiagnosed Disease Test?
The TruGenome Undiagnosed Disease Test is intended to aid in the diagnosis of inherited diseases of single-gene etiology. Using NGS technology, the Undiagnosed Disease Test provides a comprehensive view into the entire genome.

In the TruGenome Undiagnosed Disease Test, whole-genome sequencing may be performed on the patient alone, or as a trio that includes the patient and his/her parents. Trio testing can confirm whether observed variants are new in the child or inherited from one or both parents. This information can be used to refine search parameters, focusing the search for the causative variant.

The Undiagnosed Disease Test is most appropriate for situations in which evaluating genetic markers may clarify or refine the diagnosis. Examples include mitochondrial conditions, inherited cardiomyopathies, conditions involving multiple symptoms such as seizures, organ involvement, and dysmorphology, and degenerative conditions. This test may also be appropriate for evaluating inherited cancer predispositions, but is not intended to identify somatic cancer variants for guiding therapeutic choices. A complete description of the TruGenome Undiagnosed Disease Test and its intended use is available at www.illumina.com/test_descriptions.

How to get a TruGenome Undiagnosed Disease Test for Your Patient*

Discuss whole-genome sequencing with your patient.

Request a consultation with Illumina to relay pertinent medical information and select test targets.

Obtain a blood sample from your patient and send it to Illumina.

Illumina will sequence the DNA and interpret the selected genes.

Illumina sends you a report containing clinical classifications of found variants.

Meet with your patient to discuss test results and next steps in care.

What information do I receive from this test?

Data from whole-genome sequencing performed through the TruGenome Undiagnosed Disease Test is reviewed and analyzed by an in-house panel of Ph.D. geneticists, genetic counselors, and certified medical geneticists. The team provides an interpreted clinical report containing an assessment of the genes targeted based on association with the patient's phenotype. The clinical report includes an interpretation of genomic findings, details on clinically significant findings and the associated conditions, and references from which clinical associations were drawn.

Regardless of the genes targeted, an incidental findings report is supplied. This report provides screening results for genes recommended by the America College of Medical Genetics (ACMG) in the "Recommendations for Reporting of Incidental Findings in Clinical and Genome Sequencing"[3]. If trio testing is performed, a report for the parents, as well as the patient, is provided.

What is involved in getting a TruGenome Undiagnosed Disease Test for my patient?

The TruGenome Undiagnosed Disease Test is a medical test and must be ordered by a licensed physician. If your patient presents with a condition that you decide warrants a whole-genome sequencing test, start by discussing the potential value of the test with your patient or the patient's family. Within the sample collection kit, you will find a patient consent form; use this to counsel and obtain consent from your patient. You may request a consultation with an Illumina genetic counselor or medical geneticist to relay pertinent medical information and select genes that are appropriate for the test. Note that Illumina genetic counselors are not intended to support patient counseling, but to aid physicians in selecting and ordering the proper test. Using the Sample Collection Kit from Illumina, obtain a blood sample from your patient. Send the sample to the Illumina Clinical Services Laboratory where the DNA is extracted and sequenced by expert scientists using the proven Illumina NGS technology.

A team of genetic counselors, geneticists, and medical geneticists then use a series of advanced bioinformatic tools to interpret the data associated with the selected genes and generate a report containing information on the clinical classifications of any found variants. This report will be sent directly to you. Schedule time with your patient to review the test results and assess the next steps in his/her care.

About the Illumina Clinical Services Laboratory

The CLIA-certified, CAP-accredited Illumina Clinical Services Laboratory was established in 2009 for the purposes of offering human whole-genome sequencing services to physicians and genetic counselors. It is staffed with certified Clinical Laboratory Scientists who have extensive training and expertise with Illumina NGS technology. Our team of Ph.D. geneticists and certified medical geneticists has over 50 years of combined experience analyzing human genetic data.

About Illumina NGS technology

Illumina sequencing by synthesis (SBS) technology is the most published NGS technology. Using a proprietary method, DNA sequence is detected in a base-by-base manner for highly accurate, reproducible sequencing results. Learn more about sequencing at www.illumina.com/sequencing.

Learn more

To learn more about the Illumina Clinical Services Laboratory and the TruGenome Undiagnosed Disease Test, visit www.illumina.com/clinicallab.

* This graphic provides a high-level overview of the process for ordering the TruGenome Undiagnosed Disease Test from Illumina. For complete details, visit www.illumina.com/clinicallab.

References:
1. Ayme S, Rodwell C, eds (2012) 2012 Report on the state of the art of rare disease activities in Europe of the European Union Committee of Experts on Rare Disease.
2. Ng SB, Bingham AW, Buckingham KJ, Hannibal MC, McMillin MJ, et al. (2010) Exome sequencing identifies MLL2 mutations as a cause of Kabuki syndrome. Nat Genet 42: 790–793.
3. Green RC, Berg JS, Grody WW, Kalia SS, Korf BR, et al. (2013) ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. Genet Med 15: 565–574.